

A polymorphic DNA sequence from the terminal part of chromosome 20q [D20S25]

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Source/Description: The probe IP20K061 is a 0.8 kb HpaII fragment isolated from a λ phage obtained by screening a human genomic library with a subtelomeric interspersed repeat (STIR) (1). The insert is subcloned in the pBS vector (Stratagene) and can be excised with HindIII.

Polymorphism: MspI identifies 2 allelic fragments of 0.85 kb and 0.6 kb.

Frequency: Estimated from 46 parents of the CEPH panel.
0.85 kb allele: 0.55
0.60 kb allele: 0.45

Not Polymorphic For: BamHI, BglII, EcoRI, PstI, RsaI, TaqI, XbaI (studied in 5 unrelated individuals).

Chromosomal Localization: Localized to 20q13.3 by in situ hybridization and genetic linkage analysis (1).

Mendelian Inheritance: Codominant segregation observed in 19 CEPH families.

Probe Availability: The probe will be submitted to the American Tissue Culture Collection (ATCC). Until it is made available through the ATCC it can be obtained from J. Weissenbach.

Other Comments: Segregation data included in the CEPH database version 3. RFLPs are observed under normal hybridization and wash ($0.2 \times \text{SSC}$) stringencies.

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Reference: (1) Rouyer, F. *et al.* (1990) *EMBO J.* 9, 2: 505–514.

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Identification of an RsaI RFLP at the acid alpha glucosidase (GAA) locus

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Source/Description: A 1.9 kbp cDNA EcoRI segment subcloned into pUC 19. (GAA67; '2kb') (1) and subfragment indicated below.

Polymorphism: RsaI detects a two allele polymorphism 3.9 kb (A1) and 3.7 kb (A2) (also detected by a 0.7 NcoI/SacI subfragment, bp 2520 to 3236 (2))

Frequency: Studied for RsaI alleles in 35 unrelated Caucasians. Gene frequency of A1 = 0.21 and of A2 = 0.79, heterozygosity = 0.38. Fits Hardy-Weinberg equilibrium.

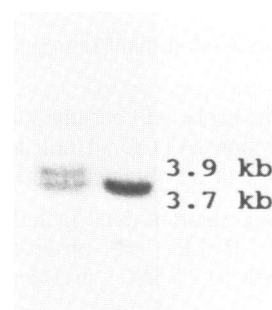
Mendelian Inheritance: Codominant inheritance in five families.

Chromosome Localization: 17q23.

Probe Availability: Available from F. Martiniuk, NYU Medical Center.

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References: (1) Martiniuk *et al.* (1986) *Proc. Natl. Acad. Sci. USA* 83, 9641–9644. (2) Martiniuk *et al.* (1990) *DNA* in press.



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